

Applicant: Lo et al.
Application No.: 09/872,063

comprising:

obtaining a serum or plasma sample from a pregnant female;
employing a primer/probe set specific for nucleic acid of interest, associated with a genetic trait, condition or abnormality not present in the pregnant female; and
amplifying the specific nucleic acid in the sample using the primer to enhance detection using the probe; and thereby
identifying the presence in the sample of the nucleic acid of fetal origin.

38. The method of claim 37 wherein the specific nucleic acid of interest is determined to differ qualitatively from that of the maternal genome of the pregnant female by comparison with a maternal nucleic acid sample from the pregnant female free of contamination by fetal nucleic acids.

39. A method performed on a serum or plasma sample taken from a pregnant female for identifying the fetal origin of a genetic trait, condition or abnormality comprising:

obtaining a serum or plasma sample from a pregnant female after the seventh week of pregnancy;
employing a probe specific for nucleic acid of interest, associated with a genetic trait, condition or abnormality not present in the pregnant female, to detect and identify the presence in the sample of the nucleic acid of fetal origin.

40. The method of claim 39 wherein the specific nucleic acid of interest is determined to differ qualitatively from that of the maternal genome of the pregnant female by comparison with a maternal nucleic acid sample from the pregnant female free of contamination by fetal nucleic acids.

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41. A method performed on a serum or plasma sample taken from a pregnant female for identifying the fetal origin of a genetic trait, condition or abnormality comprising:

obtaining a serum or plasma sample from a pregnant female;

employing a primer/probe set specific for marker nucleic acid of interest, associated with a genetic trait, condition or abnormality not present in the pregnant female;

amplifying the specific marker nucleic acid in the sample using the primer to enhance detection using the probe;

quantifying the level of marker nucleic acid present in the sample; and

determining that the level of marker nucleic acid in the sample differs from the level of marker nucleic acid in comparable maternal serum or plasma containing fetal and maternal marker nucleic acid but free of the genetic trait, condition or abnormality; and thereby

identifying the presence in the sample of the specific genetic trait, condition or abnormality of fetal origin.

42. The method of claim 41 wherein the quantification of the level of marker nucleic acid in the maternal serum or plasma is by quantitative polymerase chain reaction.

43. The method of claim 41 wherein the specific genetic trait, condition or abnormality is caused by a fetal chromosomal aneuploidy.

44. The method of claim 43 wherein the specific genetic trait, condition or

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abnormality caused by a fetal chromosomal aneuploidy is Down's Syndrome.

45. A method performed on a serum or plasma sample taken from a pregnant female for identifying the fetal origin of a genetic trait, condition or abnormality comprising:

obtaining a serum or plasma sample from a pregnant female after the seventh week of pregnancy;

employing a probe specific for marker nucleic acid of interest, associated with a genetic trait, condition or abnormality not present in the pregnant female, to detect and quantify the level of marker nucleic acid present in the sample; and

determining that the level of marker nucleic acid in the sample differs from the level of marker nucleic acid in comparable maternal serum or plasma containing fetal and maternal marker nucleic acid but free of the genetic trait, condition or abnormality to thereby identify the presence in the sample of the specific genetic trait, condition or abnormality of fetal origin.

46. A method performed on a serum or plasma sample taken from a pregnant female for identifying the fetal presence of an aneuploidal trait, condition or abnormality comprising:

obtaining a serum or plasma sample from a pregnant female after the seventh week of pregnancy;

employing a first probe specific for a chromosomal marker nucleic acid of interest, associated with a first chromosome responsible for an aneuploidal trait, condition or abnormality not present in the pregnant female, to detect and quantify the level of marker nucleic acid present in the sample;

employing a second probe specific for a second chromosomal marker nucleic acid